

WE CLAIM:

1. A method to aid in detecting the presence of tumor cells in a patient,
comprising:

5 step for determining the presence of a single basepair mutation in a
mitochondrial genome of a cell sample of a patient, wherein the mutation is found
in a tumor of the patient but not in normal tissue of the patient; and

 identifying the patient as having a tumor if one or more single
basepair mutations are determined in the mitochondrial genome of the cell sample
of the patient.

- 10 2. The method of claim 1 wherein, prior to the step for determining, the
mutation has been identified in a tumor.

3. The method of claim 1 wherein the cell sample is from a tissue suspected of
harboring a metastasis.

4. The method of claim 1 wherein the cell sample is from blood.

- 15 5. The method of claim 1 wherein the cell sample is from urine.

6. The method of claim 1 wherein the cell sample is from sputum.

7. The method of claim 1 wherein the cell sample is from saliva.

8. The method of claim 1 wherein the cell sample is from feces.

9. The method of claim 1 wherein the step for determining comprises amplifying mitochondrial DNA.

10. The method of claim 1 wherein the step for determining comprises sequencing mitochondrial DNA.

11. The method of claim 1 wherein the step for determining comprises hybridization of DNA amplified from the mitochondrial genome of the cell sample to an array of oligonucleotides which comprises matched and mismatched sequences to human mitochondrial genomic DNA.

12. The method of claim 1 wherein the single basepair mutation is a substitution mutation.

13. The method of claim 1 wherein the single basepair mutation is a one basepair insertion.

14. The method of claim 1 wherein the single basepair mutation is a one basepair deletion.

15. The method of claim 1 wherein the single basepair mutation is a transition

mutation.

16. The method of claim 1 wherein the single basepair mutation is a homoplasmic mutation.

17. The method of claim 1 wherein the single basepair mutation is a T→C substitution at position 710.

18. The method of claim 1 wherein the single basepair mutation is a T→C substitution at position 1738.

19. The method of claim 1 wherein the single basepair mutation is a T→C substitution at position 3308.

20. The method of claim 1 wherein the single basepair mutation is a G→A substitution at position 8009.

21. The method of claim 1 wherein the single basepair mutation is a G→A substitution at position 14985.

22. The method of claim 1 wherein the single basepair mutation is a T→C substitution at position 15572.

23. The method of claim 1 wherein the single basepair mutation is a G→A substitution at position 9949.

24. The method of claim 1 wherein the single basepair mutation is a T→C substitution at position 10563.

5 25. The method of claim 1 wherein the single basepair mutation is a G→A substitution at position 6264.

26. The method of claim 1 wherein the single basepair mutation is an A insertion at position 12418.

10 27. The method of claim 1 wherein the single basepair mutation is a T→C substitution at position 1967.

28. The method of claim 1 wherein the single basepair mutation is a T→A substitution at position 2299.

29. The method of claim 2 wherein the mutation was identified previously in a tumor of the patient.

15 30. The method of claim 29 wherein the patient has received anti-cancer therapy and the step for determining is performed at least three times to monitor progress

of the anti-cancer therapy.

31. The method of claim 1 further comprising a step for testing a normal tissue of the patient to determine the absence of the mutation.